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Newborn Blood Spot Card Screening

Why is my baby screened?

A small spot of your baby's blood can be used to get important information about their health. A newborn baby can look healthy but have a rare and serious disorder that you and your doctor or midwife may not know about. Newborn screening finds babies who may have one of these rare disorders. Finding and treating these disorders early prevents or reduces serious health problems. If not treated, many of these disorders can cause severe developmental delay, other health problems and even sudden infant death.

In B.C., there are about 60 babies born each year (1 out of every 750) who are found to have one of these disorders.

What will my baby be screened for?

Your baby will be screened for 27 treatable disorders. These include:

Metabolic disorders: These occur when your baby's body can't break down (metabolize) certain substances in food like fats, proteins and/or sugars. These substances can build up and cause serious health problems. Early treatment can usually prevent these problems.

Endocrine disorders: Endocrine disorders are the most common group of conditions found through a newborn blood spot screening panel. These disorders occur when your baby's thyroid or adrenal glands do not produce enough hormones.Replacing these hormones can prevent:

- Growth problems
- Intellectual disability or developmental delay
- Shock
- Unexpected death

Blood disorders: Blood disorders happen when the part of the red blood cell that carries oxygen (hemoglobin) throughout the body is changed. Hemoglobin is important because it picks up oxygen in the lungs and carries it to the other parts of the body. Serious health problems can be prevented through medicines and special treatments.

Cystic Fibrosis: Cystic fibrosis (CF) is an inherited disorder that causes thick mucus to build up in the lungs and may affect the digestive system. If your baby has CF, they may have frequent chest infections and problems digesting food. which can result in slower weight gain. Treatment can help with growth and reduce the risk of lung infections.

Spinal Muscular Atrophy (SMA): SMA affects the nerves that control muscle movement. Babies and children with SMA may have difficulty with swallowing, crawling, sitting, or walking. Treatment can slow or even stop some of these problems.

Severe Combined Immunodeficiency (SCID): SCID affects a baby's immune system and makes it hard to fight against infections or germs. Early treatment can greatly improve overall health.

For more information about these disorders and the benefits of early detection and treatment, visit Perinatal Services BC www.perinatalservicesbc.ca/our-services/screening-programs/prenatal-genetic-screening

How is my baby screened?

Your baby's heel is pricked and a few drops of blood are taken and put onto a special card. Your baby may cry, but taking the blood sample does not harm your baby. You can help comfort your baby by soothing them with your voice and providing comforting touch. In some instances you may also be able to hold and breast/chest feed them while the blood is being taken. The blood sample is sent to the laboratory at BC Children's hospital for testing. The same blood sample is used to screen for all the disorders on the screening panel.

How soon after birth will my baby be screened?

The ideal time to collect the newborn screening blood sample is between 24 and 48 hours after birth. This will be done before your baby leaves the hospital, or by your midwife at home if you have a home birth.

What if I go home with my baby less than 24 hours after birth?

A blood sample will still be taken in the hospital before leaving unless arrangements have been made with your midwife for collection in the community between 24-48 hours of life. Screening detects over 80% of disorders even at less than 24 hours of age. Early detection is important if your child has one of these disorders. You will get instructions on how to have the sample repeated within 2 weeks. The purpose of the second sample is to double check the few disorders that can be missed on the first (early) screen.

Can I wait and have my babyscreened later?

The earlier these treatable disorders are found, the better the outcome for babies with these disorders. It is strongly advised that your baby have a blood sample taken before leaving the hospital. If you decide you do not want your baby to have a blood sample taken before they leave the hospital, you will be asked to sign a form. This form shows you understand the reasons for the test and the possible outcomes for your baby if your baby is tested too late or not at all and has one of these disorders.

How do I find out the results of the screening?

Your baby's screening results are reported to your baby's doctor or midwife.

What does it mean if the screen is negative?

A negative screen means that the chance that your baby has one of these disorders is very low. Very rarely, screening may miss a baby with one of these disorders.

What does it mean if the screen is positive and what happens next?

A positive screen means that there might be a health problem. It does not mean that your baby has one of these disorders, but it is possible. More tests are needed to find out for sure. The type of follow up testing will depend on the disorders being investigated and can include more bloodwork.

If your baby has one of these disorders, you will be referred to a specialist with experience in treating these disorders.

What happens to my baby's blood sample (blood spot card) when the screening is complete?

Your baby's card with the leftover blood will be kept for 10 years in secure storage. Occasionally, the dried blood spot samples may be used for other purposes after the screening is finished. These include:

- Re-running a test if the first test result was not clear
- Trying to find the reason for a health condition that has developed later in a child's life or trying to find the cause of an unexplained illness or death of a child
- Checking the quality of testing done by the laboratory to make sure that results are accurate
- Developing better or new tests for screening of disorders

Samples may also be used for health research if the research has been approved by a Clinical Research Ethics Board. In these cases, all information that may identify the baby is removed.

If you do not want your baby's stored blood spot card to be used for these purposes, you can fill out a form called a "Directive to Destroy Leftover Newborn Screening Blood samples" and send it to the BC Newborn Screening Program. Visit www.perinatalservicesbc.ca/our-services/screening-programs/prenatal-genetic-screening for details.